CLAIMS

1. A nucleic acid sequence comprising :

1) the sequence represented in Figure 8; or

2) the sequence represented in Figure 2; or

3) a part of the sequence of Figure 2 with the proviso that it is able to code for a protein having a calcium dependant protease activity involved in a LGMD2 disease ; or

4) a sequence derived from a sequence defined in 1), 2) or 3) by substitution, deletion or addition of one or more nucleotides with the proviso that said sequence still codes for said protease.

2. A nucleic acid sequence that is complementary to a nucleic acid sequence according to claim 1.

3. A nucleic acid sequence comprising in its structure a nucleotidic sequence according to claim 1 ~~or 2~~, under the control of regulatory elements, and involved in the expression of calpaïn activity in a LGMD2 disease.

4. A nucleic acid sequence encoding the aminoacid sequence represented in Figure 2.

5. An amino acid sequence which is coded by a nucleic acid sequence according to ~~claims 1 to 4~~ claim 1, characterized in that it is a calcium dependent protease enzyme belonging to the calpaïn family, involved in the etiology of LGMD2.

6. An aminoacid sequence according to claim 5 ~~or 6~~, characterized in that either it contains the sequence such as represented in Figure 2, or the amino acid sequence of Figure 2 modified by deletion, insertion and/or replacement of one or more amino acids with the proviso that such aminoacid sequence has the calpaïn activity involved in LGMD2 disease.

7. An amino acid sequence according to claim 5 ~~or 6~~, characterized in that LGMD2 is LGMD2A.

8. A host cell unable to express a calpaïn enzyme activity, characterized in that it is transformed or transfected with a nucleic acid sequence comprising all or part of the nucleic acid sequence according to ~~any one of claims 1 to 4~~ claim 1.

9. Use of a nucleic acid according to one of claims 1 to 4 or a host cell according to claim 8 in the manufacturing of a drug for the prevention or the treatment of an LGMD2 disease.

10. Use of an amino acid sequence according to ~~claims 5 to 6~~ claim 5 in the manufacturing of a drug for the prevention or the treatment of an LGMD2 disease.

11. Use according to ~~claims 10 or 11~~ claim 10, characterized in that LGMD2 is LGMD2A.

12. Use of an amino acid sequence according to ~~claims 5 to 7~~ claim 5 for the screening of the ligands of said amino acid sequence, said ligand being selected in a group consisting of substrate(s), co-factors or regulatory components.

13. Use of a nucleic acid sequence according to ~~one of claims 1 to 4~~ claim 1 in a screening method for the determination of the components which may act on the regulation of gene expression of calpaïn.

14. Use of an host cell according to claim 8 in a screening method for the determination of components active on the expression of the calpaïn.

15. A method for detecting of a predisposition to a LGMD2 disease in a family or a human being, such method comprising the steps of :

- selecting one or more exons or their flanking sequences of the gene,
- selecting primers specific for these exons, or their flanking sequences, or an hybrid thereof,
- amplifying the nucleic acid sequences with these primers, the substrate for this amplification being the DNA of a human being; and
- comparing the amplified sequence to the corresponding sequence derived from Figure 2 or Figure 8.

16. The method according to claim 15, characterized in that the primers are those selected from the group of :

a) those described in Table 1;

b) those described in Table 3; and

c) those including the introns-exons junctions of Table 2;

d) those derived from the primers in a), b), or c).

17. The method according to claim 15 ~~or 16~~, characterized in that LGMD2 is LGMD2A.

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18. A kit for the detection of a predisposition to LGMD2 by nucleic and amplification characterized in that it comprises primers selected from the group of :

a) those described in Table 1;

b) those described in Table 3; and

c) those including the introns-exons junctions of Table 2;

d) those derived from the primers in a), b) or c).

19. Use of a host cell according to claim 8 in a manufacturing of a drug for gene therapy of an LGMD2 disease.

20. Pharmaceutical composition for the treatment of an LGMD2 disease characterized in that in contains a component selected from the group of :

a) a nucleic acid sequence according to claims 1 to 4,

b) a host cell according to claim 8,

c) an aminoacid sequence according to claims 5 to 7.